

Age-related macular degeneration

Description

Age-related macular degeneration is an eye disease that is a leading cause of vision loss in older people in developed countries. Subtle abnormalities indicating changes in vision may occur in a person's forties or fifties. Distorted vision and vision loss usually become noticeable in a person's sixties or seventies and tend to worsen over time.

Age-related macular degeneration mainly affects central vision, which is needed for detailed tasks such as reading, driving, and recognizing faces. The vision loss in this condition results from a gradual deterioration of light-sensing cells in the tissue at the back of the eye that detects light and color (the retina). Specifically, age-related macular degeneration affects a small area near the center of the retina, called the macula, which is responsible for central vision. Side (peripheral) vision and night vision are generally not affected, but slow adjustment of vision to darkness (dark adaptation) and reduced dim light (scotopic) vision often occur in the early stages of the disease.

Researchers have described two major types of age-related macular degeneration, known as the dry form and the wet form. The dry form is much more common, accounting for 85 to 90 percent of all cases of age-related macular degeneration. It is characterized by a buildup of yellowish deposits called drusen beneath the retina and vision loss that worsens slowly over time. The most advanced stage of dry age-related macular degeneration is known as geographic atrophy, in which areas of the macula waste away (atrophy), resulting in severe vision loss. Dry age-related macular degeneration typically affects vision in both eyes, although vision loss often occurs in one eye before the other.

In 10 to 15 percent of affected individuals, the dry form progresses to the wet form of age-related macular degeneration. The wet form is characterized by the growth of abnormal, fragile blood vessels underneath the macula. These vessels leak blood and fluid, which damages the macula and makes central vision appear blurry and distorted. The wet form of age-related macular degeneration is associated with severe vision loss that can worsen rapidly.

Frequency

It is estimated that 8 percent of people around the world have signs of age-related macular degeneration. The condition currently affects about 11 million Americans and 170 million people worldwide, and the prevalence is expected to increase over the

coming decades as the proportion of older people in the population increases.

For reasons that are unclear, age-related macular degeneration affects individuals of European descent more frequently than African Americans in the United States.

Causes

Age-related macular degeneration results from a combination of genetic and environmental factors. Many of these factors have been identified, but some remain unknown.

Researchers have considered changes in many genes as possible risk factors for age-related macular degeneration. The best-studied of these genes are involved in a part of the body's immune response known as the complement system. This system is a group of proteins that work together to destroy foreign invaders (such as bacteria and viruses), trigger inflammation, and remove debris from cells and tissues. Genetic changes in and around several complement system genes, including the *CFH* gene, contribute to a person's risk of developing age-related macular degeneration. It is unclear how these genetic changes are related to the retinal damage and vision loss characteristic of this condition.

Changes on the long (q) arm of chromosome 10 in a region known as 10q26 are also associated with an increased risk of age-related macular degeneration. The 10q26 region contains two genes of interest, *ARMS2* and *HTRA1*. Changes in both genes have been studied as possible risk factors for the disease. However, because the two genes are so close together, it is difficult to tell which gene is associated with age-related macular degeneration risk, or whether increased risk results from variations in both genes.

Other genes that are associated with age-related macular degeneration include genes involved in transporting and processing high-density lipoproteins (HDL, also known as "good" cholesterol) and genes that have been associated with other forms of macular disease.

Researchers have also examined nongenetic factors that contribute to the risk of age-related macular degeneration. Age appears to be the most important risk factor; the chance of developing the condition increases significantly as a person gets older. Smoking is another established risk factor for age-related macular degeneration. Other factors that may increase the risk of this condition include high blood pressure; heart disease; a diet that is high in fat, high in easily digested foods (high glycemic index), or low in certain nutrients (such as antioxidants and zinc); obesity; and exposure to ultraviolet (UV) rays from sunlight. However, it is unclear how these factors influence the risk of developing age-related macular degeneration.

[Learn more about the genes associated with Age-related macular degeneration](#)

- ABCA4
- APOE

- ARMS2
- ASPM
- BEST1
- C2
- C3
- CFH
- CFHR5
- CFI
- CST3
- ELOVL4
- ERCC6
- F13B
- FBLN5
- HTRA1
- LIPC

Additional Information from NCBI Gene:

- C9
- CETP
- CFB
- CFHR1
- CFHR2
- CFHR3
- CFHR4
- COL10A1
- COL8A1
- CX3CR1
- FILIP1L
- FRK
- HMCN1
- MAP2
- TIMP3
- TNFRSF10A
- VEGFA

Inheritance

Age-related macular degeneration usually does not have a clear-cut pattern of inheritance, although the condition appears to run in families in some cases. An estimated 15 to 20 percent of people with age-related macular degeneration have at least one first-degree relative (such as a sibling or parent) with the condition.

Other Names for This Condition

- Age-related maculopathy
- AMD
- ARMD
- Macular degeneration, age-related

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Age-related macular degeneration (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0242383/>)
- Genetic Testing Registry: Age-related macular degeneration 1 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864205/>)
- Genetic Testing Registry: Age-related macular degeneration 10 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1969108/>)
- Genetic Testing Registry: Age-related macular degeneration 11 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2677774/>)
- Genetic Testing Registry: Age-related macular degeneration 12 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151079/>)
- Genetic Testing Registry: Age-related macular degeneration 13 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3809523/>)
- Genetic Testing Registry: Age-related macular degeneration 14 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3809653/>)
- Genetic Testing Registry: Age-related macular degeneration 2 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3495438/>)
- Genetic Testing Registry: Age-related macular degeneration 3 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C4225406/>)
- Genetic Testing Registry: Age-related macular degeneration 4 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853147/>)
- Genetic Testing Registry: Age-related macular degeneration 5 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151063/>)
- Genetic Testing Registry: Age-related macular degeneration 6 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151060/>)

- Genetic Testing Registry: Age-related macular degeneration 7 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857813/>)
- Genetic Testing Registry: Age-related macular degeneration 8 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151070/>)
- Genetic Testing Registry: Age-related macular degeneration 9 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1969651/>)
- Genetic Testing Registry: Macular degeneration, age-related, 15 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3810042/>)

Genetic and Rare Diseases Information Center

- Macular degeneration (<https://rarediseases.info.nih.gov/diseases/10260/macular-degeneration>)

Patient Support and Advocacy Resources

- Disease InfoSearch (<https://www.diseaseinfosearch.org/>)
- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov (<https://clinicaltrials.gov/ct2/results?cond=%22age-related+macular+degeneration%22>)

Catalog of Genes and Diseases from OMIM

- MACULAR DEGENERATION, AGE-RELATED, 1 (<https://omim.org/entry/603075>)
- MACULAR DEGENERATION, AGE-RELATED, 10 (<https://omim.org/entry/611488>)
- MACULAR DEGENERATION, AGE-RELATED, 11 (<https://omim.org/entry/611953>)
- MACULAR DEGENERATION, AGE-RELATED, 12 (<https://omim.org/entry/613784>)
- MACULAR DEGENERATION, AGE-RELATED, 13 (<https://omim.org/entry/615439>)
- MACULAR DEGENERATION, AGE-RELATED, 14 (<https://omim.org/entry/615489>)
- MACULAR DEGENERATION, AGE-RELATED, 15 (<https://omim.org/entry/615591>)
- MACULAR DEGENERATION, AGE-RELATED, 2 (<https://omim.org/entry/153800>)
- MACULAR DEGENERATION, AGE-RELATED, 4 (<https://omim.org/entry/610698>)
- MACULAR DEGENERATION, AGE-RELATED, 5 (<https://omim.org/entry/613761>)
- MACULAR DEGENERATION, AGE-RELATED, 6 (<https://omim.org/entry/613757>)
- MACULAR DEGENERATION, AGE-RELATED, 7 (<https://omim.org/entry/610149>)
- MACULAR DEGENERATION, AGE-RELATED, 8 (<https://omim.org/entry/613778>)
- MACULAR DEGENERATION, AGE-RELATED, 9 (<https://omim.org/entry/611378>)

- NEUROPATHY, HEREDITARY, WITH OR WITHOUT AGE-RELATED MACULAR DEGENERATION (<https://omim.org/entry/608895>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Macular+Degeneration%5BM+AJR%5D%29+AND+%28age-related+macular+degeneration%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>)

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Last updated October 4, 2021